

Supplementary Table 1: Clinical data of both cases with *C2CD3* mutations

Case	1	2
Sex	Male	Male
Age	4 years	22 wg
<i>OFD features</i>		
Facial dysmorphism	+	+
Cleft tongue	+	-
Lobulated tongue	+	-
Buccal frenulae	+	+
Lingual hamartoma	+	-
Cleft palate	+	-
Absent epiglottis	+	-
Supernumerary teeth	+	NA
Hand post axial polydactyly	Bilateral	Bilateral
Broad hallux	Bilateral	Bilateral
<i>Microcephaly</i>	OFC 42.3cm (-5 SD)	brain weight 50g (-4 SD)
<i>Neurological and sensorial features</i>		
Ataxia	-	NA
Abnormal eye movements	-	NA
Nystagmus	-	NA
Ventilatory disorders (apnea, hyperpnea)	-	NA
Deafness	-	NA
Retinopathy	+	NA
Intellectual disability	+*	NA
<i>Brain malformations</i>		
Vermian hypoplasia	MTS	+
Incomplete corpus callosum agenesis	+	+
Subarachnoid cysts	+	+
Myelinisation defect	+	-
<i>Other malformations</i>		
Cardiac	-	-
Abdominorenal	-	Right renal hypoplasia Micropenis
Genital	Micropenis	

MTS: Molar Tooth Sign, NA: not available, SD: standard deviation, wg : weeks of gestation, * walking but no speech

Supplementary Table 2. Antibodies used in this study, Related to Online Methods

Antigen	Source	Application	Reference
OFD1 (human)	Andrew Fry	IB, IP	21
Odf1	Brunella Franco	IF	27
Sass6	Laurence Pelletier	IF	32
Polyglutamylated tubulin	mAb GT335; Carsten Janke	IF	45
Ninein	Michel Bornens	IF	46
Cep164	Tim Stearns	IF	47
Centrin	mAb 20H5 (Millipore)	IF	48
Centrin	Sigma (C7736)	IB	
PCM-1	Andreas Merdes	IF	17
CPAP	Proteintech (11517-1-AP)	IF	
IFT88	Proteintech (13967-1-AP)	IF	
CP110	Proteintech (12780-1-AP)	IF	
C2CD3 (human)	Sigma (HPA038552)	IB	
Odf2	Sigma (HPA001874)	IF	
Alpha Tubulin	mAb DM1A (Thermo)	IF	
Tyrosinated alpha Tubulin	mAb YL1/2 (Serotec)	IF	
Gamma Tubulin	mAb GTU88 (Sigma)	IF	
Cep290	Sophie Saunier	IF	49
GFP	Maxence Nachury	IB	19
Myc	mAb 9E10 (Covance)	IB, IP	

IF: Immunofluorescence, IB: Immunoblotting, IP: Immunoprecipitation, mAb: monoclonal antibody

References for Supplementary Material

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